MC2R gene

melanocortin 2 receptor

Normal Function

The *MC2R* gene provides instructions for making a protein called adrenocorticotropic hormone (ACTH) receptor. This protein is found primarily in the adrenal glands, which are hormone-producing glands located on top of each kidney. The ACTH receptor is embedded in the membrane of cells where it attaches (binds) to ACTH. ACTH is a hormone that is released by the pituitary gland, located at the base of the brain. The binding of ACTH to its receptor triggers the adrenal glands to produce a group of hormones called glucocorticoids. These hormones, which include cortisol and corticosterone, aid in immune system function, play a role in maintaining normal blood sugar levels, help trigger nerve cell signaling in the brain, and serve many other purposes in the body.

The ACTH receptor also likely plays a role in the development of the adrenal glands before birth.

Health Conditions Related to Genetic Changes

familial glucocorticoid deficiency

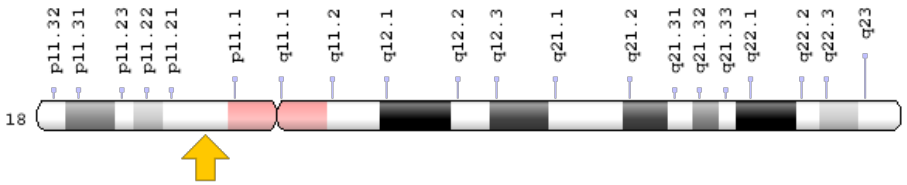
More than 40 mutations in the *MC2R* gene have been found to cause familial glucocorticoid deficiency. This condition is characterized by potentially life-threatening low blood sugar (hypoglycemia), recurrent infections, and skin coloring darker than that of other family members (hyperpigmentation). *MC2R* gene mutations account for approximately 25 percent of cases of this condition. Most of these mutations change single protein building blocks (amino acids) in the ACTH receptor. As a result, the receptor cannot be transported to the cell membrane or bind to ACTH. Without the binding of the ACTH receptor to its hormone, there is no signal to trigger the adrenal glands to produce glucocorticoids. A shortage of these hormones impairs blood sugar regulation, immune system function, and other cellular functions, leading to the signs and symptoms of familial glucocorticoid deficiency.

primary macronodular adrenal hyperplasia

Chromosomal Location

Cytogenetic Location: 18p11.21, which is the short (p) arm of chromosome 18 at position 11.21

Molecular Location: base pairs 13,882,042 to 13,915,707 on chromosome 18 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ACTH receptor
- ACTHR
- ACTHR_HUMAN
- adrenocorticotropic hormone receptor
- adrenocorticotropin receptor
- corticotropin receptor
- MC2 receptor
- melanocortin 2 receptor (adrenocorticotropic hormone)

Additional Information & Resources

Educational Resources

- Endocrinology: An Integrated Approach (2001): Feedback Control of Glucocorticoids
  https://www.ncbi.nlm.nih.gov/books/NBK26/#A526

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MC2R%5BTIAB%5D%29+OR+%28melanocortin+2+receptor%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MELANOCORTIN 2 RECEPTOR
  http://omim.org/entry/607397

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MC2R.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MC2R%5Bgene%5D
- HGNC Gene Family: Melanocortin receptors
  http://www.genenames.org/cgi-bin/genefamilies/set/236
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6930
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4158
- UniProt
  http://www.uniprot.org/uniprot/Q01718

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